

## ELOVL4 gene

ELOVL fatty acid elongase 4

### Normal Function

The *ELOVL4* gene provides instructions for making a protein that is found primarily in the retina, the specialized light-sensitive tissue that lines the back of the eye.

Specifically, the ELOVL4 protein is produced in the retina's light-sensing cells (photoreceptors). The ELOVL4 protein is also found in the brain and skin, but less is known about its activity (expression) in these structures.

Inside photoreceptors, this protein is located in a cell structure called the endoplasmic reticulum that is involved in protein production, processing, and transport. The ELOVL4 protein plays a role in making a group of fats called very long-chain fatty acids. The protein helps add carbon molecules to long-chain fatty acids, making them very long-chain fatty acids. The function of the very long-chain fatty acids produced by the ELOVL4 protein is unknown.

### Health Conditions Related to Genetic Changes

#### Stargardt macular degeneration

A few variants (also called mutations) in the *ELOVL4* gene have been found to cause Stargardt macular degeneration. These variants create a premature stop signal in the instructions used to make the ELOVL4 protein. This causes the protein to form clumps (aggregates) in the endoplasmic reticulum of photoreceptors. These aggregates cannot make very long-chain fatty acids and may interfere with cell functions, ultimately leading to cell death. The loss of photoreceptors causes progressive vision loss in people with Stargardt macular degeneration.

#### Age-related macular degeneration

MedlinePlus Genetics provides information about Age-related macular degeneration

### Other Names for This Gene

- elongation of very long chain fatty acids (FEN1/Elo2, SUR4/Elo3, yeast)-like 4
- elongation of very long chain fatty acids protein 4
- ELOV4\_HUMAN

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of ELOVL4 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6785\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6785[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28ELOVL4%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D>)

### Catalog of Genes and Diseases from OMIM

- ELONGATION OF VERY LONG CHAIN FATTY ACIDS-LIKE 4; ELOVL4 (<https://omim.org/entry/605512>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6785>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ELOVL4\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ELOVL4[gene]))

## References

- Agbaga MP, Brush RS, Mandal MN, Henry K, Elliott MH, Anderson RE. Role of Stargardt-3 macular dystrophy protein (ELOVL4) in the biosynthesis of very longchain fatty acids. *Proc Natl Acad Sci U S A*. 2008 Sep 2;105(35):12843-8. doi:10.1073/pnas.0802607105. Epub 2008 Aug 26. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/18728184>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2525561/>)
- Molday RS, Zhang K. Defective lipid transport and biosynthesis in recessive and dominant Stargardt macular degeneration. *Prog Lipid Res*. 2010 Oct;49(4):476-92. doi: 10.1016/j.plipres.2010.07.002. Epub 2010 Jul 13. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/20633576>) or Free article on PubMed Central (<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2946192/>)
- Walia S, Fishman GA. Natural history of phenotypic changes in Stargardt macular dystrophy. *Ophthalmic Genet*. 2009 Jun;30(2):63-8. doi:10.1080/13816810802695550. Citation on PubMed (<https://pubmed.ncbi.nlm.nih.gov/19373676>)

## Genomic Location

The *ELOVL4* gene is found on chromosome 6 (<https://medlineplus.gov/genetics/chromosome/6/>).

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